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MINISTRY OF SCIENCE AND HIGHER EDUCATION

RUSSIAN FEDERATION

Federal State Autonomous Educational Institution of Higher Education

NORTH-CAUCASUS FEDERAL UNIVERSITY

APPROVED

Assoc. Prof. Kubanov S.I.

PhD in Medical Sciences

Dean

Faculty of Medicine and Biology

COURSE OUTLINE OF THE DISCIPLINE (MODULE)

Medical Genetics

- Specialty: 31.05.01 General Medicine
- Mode of training: Full-time
- Year of commencement of studies: 2026
- Delivered in Semester 2

Developed by:

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1. Aim and Objectives of Mastering the Discipline (Module)

- Aim of mastering the discipline «Medical Genetics» is to provide students with the necessary knowledge and understanding in the field of general and medical genetics, to equip them with practical skills and abilities for work in a genetics laboratory, to enable them to master biochemical and immunogenetic research methods, to develop skills in collecting genealogical (family) histories, identifying the hereditary nature and mode of inheritance of diseases, as well as to build students' foundational knowledge in clinical genetics, including the ability to diagnose major hereditary disorders and implement their prevention.
- Objectives of mastering the discipline «Medical Genetics»:
 1. To develop a comprehensive understanding of the importance of genetics in human life and the organization of hereditary (genetic) material.
 2. To provide thorough knowledge of the laws of heredity and variation (inheritance and variability), as well as the semiotics (clinical signs) and clinical diagnosis of hereditary diseases.
 3. To familiarize students with both traditional and modern laboratory research methods in genetics, including prenatal diagnostic techniques.
 4. To master the methods used for Medical Genetics.

2. Place of the Discipline (Module) in the Structure of the Training (educational) Program

The discipline «Medical Genetics» belongs to the part developed by the parties involved in academic relations).

3. List of Potential Learning Outcomes for the Discipline (Module), Correlated with the Outcomes of Mastering the Educational Program

Code and definition of the competence	Code and definition of the indicator	Planned learning outcomes for the discipline (module) describing the stages of competency and indicator development
PC-2. Ready to collect and analyze patient complaints, data of their medical history, examination results, laboratory, instrumental, pathological-anatomical, and other investigations to recognize a condition or establish the fact of presence or absence of a disease.	AI-1 PC-2: Knows how to collect complaints, medical history of life and disease of the patient, and analyze the information obtained. AI-2 PC-2: Conducts a complete physical examination of the patient (inspection, palpation, percussion, auscultation) and interprets its results. AI-3 PC-2: Defines a preliminary diagnosis and compiles a plan for laboratory and instrumental examinations of the patient.	Formation of clinical thinking
PC-3. Capable of determining the patient's	AI-1 PC-3: Capable of constructing an algorithm and	Acquisition of special medical skills and their

main pathological conditions, symptoms, disease syndromes, and nosological forms in accordance with the International Statistical Classification of Diseases and Related Health Problems, 10th Revision.	choosing criteria for determining the main pathological states, syndromes, and diseases in patients of various profiles in accordance with the International Statistical Classification of Diseases and Related Health Problems, 10th Revision (ICD-10). AI-2 PC-3: Identifies the main pathological symptoms and syndromes, and formulates a clinical diagnosis in accordance with ICD-10.	application in practical healthcare
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4. Volume of the Academic Discipline (Module) and Control *

Volume of classes: Total: 3 ECTS credits, 108 academic hours.	Full-time type of study, in academic hours
Face-to-face work	54
Lectures / including practical training	18/0
Practical classes / including practical training	36/0
Independent work	54
Forms of control	
Graded credit (Graded Pass/Fail test)	

* The discipline (module) provides for the use of e-learning and remote learning technologies (unless otherwise set by the educational standard).

5. Content of the Discipline (Module), Structured by Topics (Sections) with an Indication of the Hours and Types of Classes

#	Section (topic) of the discipline and brief content	Compe- tencies and indica- tors to be deve- loped	Full-time training			Forms of current control of academic progress
			Contact work of students with the teacher / of them practical skills training, hours		Independent work, hours	
			Lectures	Practical		

1	Introduction to Medical Genetics. Methods of Medical Genetics. Basic concepts of medical genetics. History of medical genetics. Axioms (fundamental principles) of medical genetics. Genomics and clinical medicine. Subject and objectives of medical genetics. The increasing proportion of hereditary pathology in morbidity and disability rates. Methods of Medical Genetics and Laboratory Diagnostic Techniques for Hereditary Diseases.	AI-1 PC-2 AI-2 PC-2 AI-3 PC-2 AI-1 PC-3 AI-2 PC-3	2		4	6	Presentation of report Testing
2	Morphogenetic variants of development. Congenital malformations (Developmental defects). The role of heredity and environment in the development of pathology. Morphogenetic variants of development: their genesis and postnatal modification. General and specific morphogenetic variants: their significance in the diagnosis of hereditary syndromes and congenital conditions. Classification of hereditary pathology. Congenital malformations: primary and secondary. Isolated, systemic, and multiple defects. Etiological heterogeneity of congenital malformations. The teratogenic termination period (critical periods of development). Mutations. Classification of mutations. The mutational process and environmental factors. Radiation mutagenesis.	AI-1 PC-2 AI-2 PC-2 AI-3 PC-2 AI-1 PC-3 AI-2 PC-3	2		4	6	Presentation of report Testing
3	Gene disorders (Monogenic diseases). General principles of pathogenesis. Etiology, classification, and clinical presentation. The concept of monogenic disease. Types of gene mutations. Classification of monogenic disorders. Nomenclature for recording mutations in human genes. Genetic heterogeneity and clinical polymorphism of monogenic diseases. Pathogenesis of monogenic disorders. Clinical and genetic characteristics of monogenic diseases with Mendelian inheritance. Autosomal dominant monogenic disorders. Hereditary motor-sensory neuropathy type 1A (Charcot–Marie–Tooth disease type 1A). Channelopathies (disorders of ion channels). Autosomal recessive monogenic disorders. Cystic fibrosis (mucoviscidosis). Proximal spinal muscular atrophy (SMA).	AI-1 PC-2 AI-2 PC-2 AI-3 PC-2 AI-1 PC-3 AI-2 PC-3	2		4	6	Presentation of report Testing

4	Chromosomal disorders (chromosomal diseases). General characteristics of chromosomal disorders. Etiology. Cytogenetics of chromosomal disorders. Pathogenesis of chromosomal disorders. Classification of chromosomal abnormalities. General clinical characteristics of chromosomal disorders. Diagnostic methods for chromosomal disorders. Therapeutic and rehabilitation options for patients.	AI-1 PC-2 AI-2 PC-2 AI-3 PC-2 AI-1 PC-3 AI-2 PC-3	2		4	6	Presentation of report Testing
5	Clinical and genetic characteristics of syndromes associated with autosomal numerical abnormalities. Classification of chromosomal disorders. Down syndrome (Trisomy 21). Etiology, pathogenesis, clinical features, and management. Edwards syndrome (Trisomy 18). Etiology, pathogenesis, clinical features, and management. Patau syndrome (Trisomy 13). Etiology, pathogenesis, clinical features, and management. Turner syndrome (Shereshevsky–Turner syndrome; 45,X). Etiology, pathogenesis, clinical features, and management.	AI-1 PC-2 AI-2 PC-2 AI-3 PC-2 AI-1 PC-3 AI-2 PC-3	2		4	6	Presentation of report Testing
6	Diseases with non-traditional inheritance patterns. Sex-linked inheritance. Duchenne and Becker muscular dystrophy (X-linked recessive). Characteristics of X-linked dominant inheritance. Holandric (Y-linked) inheritance. Mitochondrial diseases. Etiology and pathogenesis of mitochondrial disorders. Diseases of genomic imprinting. Etiology and pathogenesis of imprinting disorders. Trinucleotide repeat expansion diseases. Huntington's disease. Prion diseases. Diseases with hereditary predisposition (Multifactorial diseases).	AI-1 PC-2 AI-2 PC-2 AI-3 PC-2 AI-1 PC-3 AI-2 PC-3	2		4	6	Presentation of report Testing
7	Prevention of hereditary and congenital pathology. Genetic foundations of prevention of hereditary pathology. Types and stages of medical genetic counseling. Genetic risk (concept and calculation methods). Non-invasive and invasive prenatal diagnostic methods. Indications for invasive prenatal diagnosis. Neonatal screening.	AI-1 PC-2 AI-2 PC-2 AI-3 PC-2 AI-1 PC-3 AI-2 PC-3	2		4	6	Presentation of report Testing

8	Hereditary metabolic disorders. Genetic aspects of carcinogenesis. Metabolic disorders. General characteristics of monogenic pathology. Inherited disorders of amino acid metabolism (aminoacidopathies). Albinism. Inherited disorders of carbohydrate metabolism. Galactosemia. Glycogen storage diseases (glycogenoses). Inherited disorders of lipid metabolism. Inherited disorders of erythrocytes (red blood cell disorders). Non-spherocytic anemias. Spherocytic hemolytic anemias (hereditary spherocytosis). Lysosomal storage diseases. Mucopolysaccharidoses. Peroxisomal disorders. Stages of the genetic concept of carcinogenesis.	AI-1 PC-2 AI-2 PC-2 AI-3 PC-2 AI-1 PC-3 AI-2 PC-3	2		4	6	Presentation of report Testing
9	Prevention of hereditary pathology. Medical genetic counseling as the basis of primary prevention of hereditary diseases. Prenatal diagnosis of hereditary diseases. Preimplantation genetic diagnosis (PGD) of hereditary diseases. Biochemical screening programs as the basis of secondary prevention of hereditary pathology. Principles of treatment of hereditary diseases. Symptomatic, pathogenetic, and etiological (causal) treatment. Gene therapy. Ethical and legal issues in medical genetics. Bioethical problems of gene therapy.	AI-1 PC-2 AI-2 PC-2 AI-3 PC-2 AI-1 PC-3 AI-2 PC-3	2		4	6	Presentation of report Colloquium
	TOTAL in Semester		18		36	54	
	TOTAL		18		36	54	

6. Assessment Tools Fund for the Discipline (Module)

The assessment tools fund (ATF) for the discipline (module) «Biology: Cytology» is based on the list of competencies to be mastered, indicating the stages of their formation in the process of mastering the discipline (module). The ATF provides an objective control over the achievement of the planned learning outcomes. The ATF includes:

- A description of indicators and criteria for evaluating competencies at various stages of their formation, a description of evaluation scales;
- Methodological materials determining the procedures for evaluating knowledge, skills, abilities and (or) work experience characterizing the stages of competency formation;
- Standard control tasks and other materials necessary for assessing knowledge, skills, and the level of mastery of the formed competencies in the process of mastering the discipline (module).

The ATF is an appendix to this discipline (module) program.

7. Methodological Guidelines for Students on Mastering the Discipline

When starting work, each student must take into account the following provisions.
 The discipline (module) is structured on a thematic principle; each topic represents a logically completed section.
 The lecture material is dedicated to the consideration of key, basic provisions of the courses and the clarification of learning tasks assigned for the independent work of students.
 Laboratory works are aimed at gaining practical experience in the relevant subject area.
 The independent work of students is aimed at the independent study of additional material, preparation for practical and laboratory classes, as well as the execution of all types of independent work.
 In order to successfully master the discipline, it is important to complete all types of independent work employing the recommended sources of information.

8. Educational, Methodological, and Informational Support for the Discipline

8.1. List of Main and Additional Literature Required for Mastering the Discipline (Module)

8.1.1. List of Main Literature:

1. Genetics and genomics in medicine / Ronald Doron Cohn, Stephen W. Scherer, Ada Hamosh - Elsevier. – 2024. – 612 p.

8.1.2. List of Additional Literature:

1. Genetics: analysis & principles, Sixth edition / Robert J. Brooker - New York NY : McGraw-Hill Education. -2018. – 872 p.
2. Lewin's GENES XII / Jocelyn E. Krebs, Elliott S. Goldstein, Stephen T. Kilpatrick. - Jones & Bartlett Learning . – 2018. – 1026p.

8.2. List of Educational and Methodological Support for the Independent Work of Students in the Discipline (Module)

1. Methodological Guidelines for Students on arranging and performing independent work in the Discipline of Medical Genetics for Students majoring (Specialty) in 31.05.01 General Medicine. – Stavropol:NCFU. – 2026. – 4p.
2. Laboratory Manual for the Discipline of Medical Genetics for Students majoring (Specialty) in 31.05.01 General Medicine. – Stavropol:NCFU. – 2026. – 92p.

8.3. List of Resources of the Information and Telecommunications Network **Internet Required for Mastering the Discipline (Module)**

1. Genetics: The Fundamentals <https://mitxonline.mit.edu/courses/course-v1:MITxT+7.03.1x/>
2. National Center for Biotechnology Information <https://www.ncbi.nlm.nih.gov/>
3. Ensembl Project <https://www.ensembl.org/index.html>
4. <https://elementy.ru>

9. List of Information Technologies Employed through the Discipline (Module), Including a List of Software and Information Reference Systems

When delivering lectures, computer equipment and demonstrations of multimedia presentation materials are used. In seminars and practical classes, students deliver presentations prepared by them during the hours of independent work.

Information Reference Systems:

Information-reference and information-legal systems used in the study of the discipline:

1.	https://www.iprbookshop.ru/
2.	https://biblioclub.ru/
3.	webofknowledge.com
4.	http://scopus.com/

Software:

1.	Alt Workstation 10
2.	Alt Workstation K

3.	Alt Server
4.	Office software package – P7-Office

10. Description of the Material and Technical Resources Required for the Educational Process for the Discipline (Module)

Lecture classes	A classroom equipped with multimedia equipment and technical training facilities.
Laboratory classes	Teaching laboratory equipped with instructional equipment and technical teaching aids: a set of educational furniture with 12 student workstations, a projector, and a laptop with the necessary licensed software package. The laboratory is also equipped with the following specialized instrumentation and supplies: High-performance sequencer with a sample preparation station and a starter reagent kit. Nucleic acid amplifier with hybridization-fluorescence detection of amplification products (real-time PCR system). Single-channel variable-volume pipettors (adjustable pipettes). Pipette stand. Laboratory freezer. Liquid nitrogen storage system (liquid nitrogen tank and storage container). Thermal cycler (thermocycler). Gel documentation system with a blue light source. Horizontal electrophoresis system. Power supply for electrophoresis and blotting procedures. Spectrophotometer. Water purification system. High-speed refrigerated centrifuge. Shaking incubator. Aspirator with trap flask. Centrifuge/vortex for PCR plates. Densitometer. Refrigerated thermoshaker for microcentrifuge tubes. Magnetic stand. Vortex mixer. Magnetic stirrer with heating. Bactericidal (UV) air recirculator. Microscopes. Sets of microscope slides (prepared slides) on genetics.
Practical classes	A classroom, equipped with multimedia equipment and technical training facilities.
Independent work	A room for the independent work of students, equipped with computer hardware with the capability to connect to the Internet network and provide access to the electronic information and educational environment of the university.
Practical training	Carried out in the structural subdivisions of the university and (or) in organizations engaged in activities matching the profile of the respective training program, including its structural subdivision.

11. Specific Features of Mastering the Discipline (Module) by Persons with Disabilities

Students with disabilities shall be provided with special textbooks, teaching aids, and didactic materials, special technical training facilities for collective and individual use, the services of an assistant (helper) providing the necessary technical assistance to the students, as well as the services of sign language interpreters and tactile sign language interpreters.

The mastery of the discipline (module) by students with disabilities may be organized jointly with other students, as well as in separate groups.

Mastering the discipline (module) by students with disabilities is to be carried out taking into account the peculiarities of psychophysical development, individual capabilities, and health status.

In order to ensure the accessibility of higher education under the educational program for persons with disabilities, the following is provided during the mastery of the discipline (module):

- 1) for persons with visual impairments:
 - the presence of an assistant offering the student the necessary technical assistance, taking into account individual features (help taking the workplace, moving around, reading and formatting the task, including writing under dictation),
 - written tasks, as well as instructions on the procedure for their execution, are formatted in an enlarged font,
 - special textbooks, teaching aids, and didactic materials (having a large font or audio files),individual uniform lighting of at least 300 lux,
if necessary, a magnifying device is provided to the student to complete the task;
- 2) for persons with hearing impairments:
 - the presence of an assistant providing the student with the necessary technical assistance, taking into account individual characteristics (helps to take a workplace, move around, read and format the task, including writing with dictation),
 - the availability of sound-amplifying equipment for collective use is ensured; if necessary, sound-amplifying equipment for individual use is provided to the student;
 - provision with proper sound means of reproducing information is ensured;
- 3) for persons with musculoskeletal disorders (including severe motor function impairments of the upper limbs or the absence of upper limbs):
 - written tasks are performed on a computer with specialized software or are dictated to an assistant;
 - at the request of the student, tasks may be performed verbally.

12. Specific Features of Implementing the Discipline with the Use of Distance Learning Technologies and E-Learning

Subject to Part 1 of Article 16 of the Federal Law of December 29, 2012, No. 273-FZ On Education in the Russian Federation, e-learning shall be viewed as the arrangement of educational activities employing information contained in databases and used in the implementation of educational programs, as well as information technologies, technical means, and information and telecommunication networks that ensure the processing and transmission of the specified information via communication lines, and the interaction of students and pedagogical workers.

Remote (distance) learning technologies are understood as educational technologies implemented mainly with the use of information and telecommunication networks during the indirect (at a distance) interaction of students and pedagogical workers.

The implementation of the discipline may be carried out with distance learning technologies and e-learning in full or in part. The components of the Educational And Methodological Complex (EMC) of the discipline (working program of the discipline,

assessment and methodological materials, forms of attestation) implemented with the use of distance learning technologies and e-learning contain an indication of their use.

When arranging educational activities with distance learning technologies and e-learning, asynchronous and synchronous methods of interaction between participants in educational relations via the information and telecommunications network Internet may be offered.

When using distance learning technologies and e-learning, the schedule for the discipline shall indicate: methods of interaction among participants in educational relations via the information and telecommunications network Internet (VCC – videoconferencing, ET – electronic testing); links to the electronic information and educational environment of NCFU, to educational platforms and resources of other organizations to which open access is provided via the information and telecommunications network Internet; for synchronous learning – the time of online classes and instructors; for asynchronous learning – the authors of online courses.

When arranging intermediate attestation with distance learning technologies and e-learning, the Methodological Recommendations on the use of technical means ensuring the objective reliability of results when conducting intermediate and state final attestation for higher education educational programs – Bachelor's Degree programs, Specialist's Degree programs, and Master's Degree programs with distance learning technologies are used (Letter of the Ministry of Science and Higher Education of Russia dated December 7, 2020, # MN-19/1573-AN On sending methodological recommendations).

The implementation of the discipline with the use of e-learning and distance learning technologies is carried out using the electronic information and educational environment of NCFU, to which students are provided access via the information and telecommunications network Internet, or using the resources of other organizations, including platforms providing services for videoconferencing, online meetings, and distance learning (MTS-Link), as well as using the capabilities of social networks to carry out communication between students and instructors.

The educational and methodological support for a discipline shall be ensured with e-learning and distance learning technologies includes the working program presented in electronic form, educational and methodological manuals or a course of lectures, methodological guidelines for performing various types of educational activities of students provided for by the discipline, and other educational and methodological materials posted in the information and educational environment of NCFU.